



## MCCC2 gene

methylcrotonyl-CoA carboxylase 2

### Normal Function

The *MCCC2* gene provides instructions for making one part (the beta subunit) of an enzyme called 3-methylcrotonyl-CoA carboxylase or 3-MCC. Beta subunits join with larger alpha subunits made from the *MCCC1* gene; six of these pairings together form a functioning enzyme.

The 3-MCC enzyme is found in mitochondria, which are the energy-producing centers inside cells. This enzyme plays a critical role in breaking down proteins obtained from the diet. Specifically, it is responsible for the fourth step in the breakdown of leucine, an amino acid that is a building block of many proteins. This step converts a molecule called 3-methylcrotonyl-CoA to a molecule called 3-methylglutaconyl-CoA. Additional chemical reactions convert 3-methylglutaconyl-CoA into molecules that are later used for energy.

### Health Conditions Related to Genetic Changes

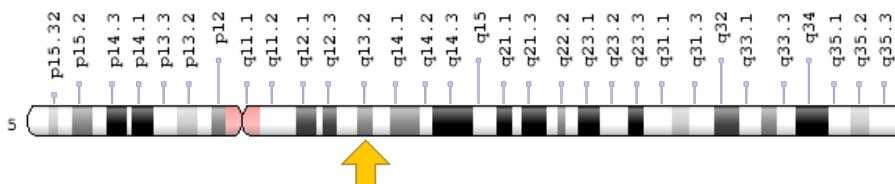
#### 3-methylcrotonyl-CoA carboxylase deficiency

More than 40 mutations in the *MCCC2* gene have been identified in people with 3-methylcrotonyl-CoA carboxylase deficiency (also known as 3-MCC deficiency). Most of these mutations change single amino acids in 3-MCC, which severely reduces the activity of the enzyme. Other mutations prevent the production of any functional enzyme. As a result, leucine cannot be broken down properly, and byproducts of leucine processing build up to toxic levels in the body. These toxic substances can damage the brain, causing the characteristic signs and symptoms of 3-MCC deficiency.

## Chromosomal Location

Cytogenetic Location: 5q13.2, which is the long (q) arm of chromosome 5 at position 13.2

Molecular Location: base pairs 71,587,288 to 71,658,706 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- 3-methylcrotonyl-CoA carboxylase non-biotin-containing subunit
- MCCase subunit beta
- MCCB
- MCCB\_HUMAN
- methylcrotonyl-CoA carboxylase 2 (beta)
- non-biotin containing subunit of 3-methylcrotonyl-CoA carboxylase

## Additional Information & Resources

### Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Major pathways of branched-chain amino acid metabolism  
<https://www.ncbi.nlm.nih.gov/books/NBK20436/figure/A3097/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28MCCC2%5BTIAB%5D%29+OR+%28MCCB%5BTIAB%5D%29+OR+%283-methylcrotonyl-CoA+carboxylase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- 3-METHYLCROTONYL-CoA CARBOXYLASE 2  
<http://omim.org/entry/609014>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=MCCC2%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=6937](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6937)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/64087>
- UniProt  
<http://www.uniprot.org/uniprot/Q9HCC0>

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Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/MCCC2>

Reviewed: October 2008

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services